



LMX1B gene

LIM homeobox transcription factor 1 beta

Normal Function

The *LMX1B* gene provides instructions for producing a protein that attaches (binds) to specific regions of DNA and regulates the activity of other genes. On the basis of this role, the LMX1B protein is called a transcription factor. The LMX1B protein appears to be particularly important during early embryonic development of the limbs, kidneys, and eyes.

Health Conditions Related to Genetic Changes

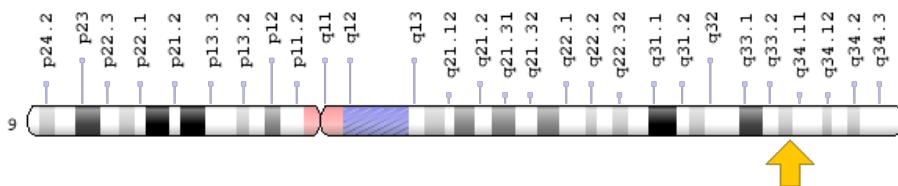
nail-patella syndrome

At least 145 mutations in the *LMX1B* gene have been found to cause nail-patella syndrome. Most mutations result in the production of an abnormally short, nonfunctional version of the LMX1B protein or change a single protein building block (amino acid). Mutations that substitute one amino acid for another amino acid reduce or eliminate the protein's ability to bind to DNA, disrupting the regulation of other genes during early development. Deletions of the entire *LMX1B* gene or large portions of the gene have also been shown to cause nail patella syndrome. It is unclear exactly how mutations in the *LMX1B* gene lead to the signs and symptoms of nail-patella syndrome.

Chromosomal Location

Cytogenetic Location: 9q33.3, which is the long (q) arm of chromosome 9 at position 33.3

Molecular Location: base pairs 126,614,443 to 126,701,032 on chromosome 9 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- LIM homeo box transcription factor 1, beta
- LIM homeobox transcription factor 1, beta
- LMX1.2
- LMX1B_HUMAN
- MGC138325
- MGC142051
- NPS1

Additional Information & Resources

Educational Resources

- Developmental Biology (sixth edition, 2000): The Generation of the Dorsal-Ventral Axis
<https://www.ncbi.nlm.nih.gov/books/NBK10053/>

GeneReviews

- Nail-Patella Syndrome
<https://www.ncbi.nlm.nih.gov/books/NBK1132>

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28LMX1B%5BTIAB%5D%29+OR+%28NPS1%5BTIAB%5D%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1800+days%22%5Bdp%5D>

OMIM

- LIM HOMEobox TRANSCRIPTION FACTOR 1, BETA
<http://omim.org/entry/602575>

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
http://atlasgeneticsoncology.org/Genes/GC_LMX1B.html
- ClinVar
<https://www.ncbi.nlm.nih.gov/clinvar?term=LMX1B%5Bgene%5D>
- HGNC Gene Family: LIM class homeoboxes
<http://www.genenames.org/cgi-bin/genefamilies/set/522>

- HGNC Gene Symbol Report
http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=6654
- NCBI Gene
<https://www.ncbi.nlm.nih.gov/gene/4010>
- UniProt
<http://www.uniprot.org/uniprot/O60663>

Sources for This Summary

- Bongers EM, Huysmans FT, Levchenko E, de Rooy JW, Blickman JG, Admiraal RJ, Huygen PL, Cruysberg JR, Toolens PA, Prins JB, Krabbe PF, Borm GF, Schoots J, van Bokhoven H, van Remortele AM, Hoefsloot LH, van Kampen A, Knoers NV. Genotype-phenotype studies in nail-patella syndrome show that LMX1B mutation location is involved in the risk of developing nephropathy. *Eur J Hum Genet.* 2005 Aug;13(8):935-46.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/15928687>
- Bongers EM, de Wijs IJ, Marcelis C, Hoefsloot LH, Knoers NV. Identification of entire LMX1B gene deletions in nail patella syndrome: evidence for haploinsufficiency as the main pathogenic mechanism underlying dominant inheritance in man. *Eur J Hum Genet.* 2008 Oct;16(10):1240-4. doi: 10.1038/ejhg.2008.83. Epub 2008 Apr 16.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/18414507>
- Dunston JA, Hamlington JD, Zaveri J, Sweeney E, Sibbring J, Tran C, Malbroux M, O'Neill JP, Mountford R, McIntosh I. The human LMX1B gene: transcription unit, promoter, and pathogenic mutations. *Genomics.* 2004 Sep;84(3):565-76.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/15498463>
- GeneReview: Nail-Patella Syndrome
<https://www.ncbi.nlm.nih.gov/books/NBK1132>
- Sato U, Kitanaka S, Sekine T, Takahashi S, Ashida A, Igarashi T. Functional characterization of LMX1B mutations associated with nail-patella syndrome. *Pediatr Res.* 2005 Jun;57(6):783-8. Epub 2005 Mar 17.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/15774843>

Reprinted from Genetics Home Reference:

<https://ghr.nlm.nih.gov/gene/LMX1B>

Reviewed: May 2008

Published: March 21, 2017

Lister Hill National Center for Biomedical Communications
U.S. National Library of Medicine
National Institutes of Health
Department of Health & Human Services